



The California Prenatal Screening Program

California Department of Public Health
Genetic Disease Screening Program

Program Information and Updates, November 2013

Prenatal Screening Program Changes, Effective November 2013

Prenatal Screening Program adds Non-invasive Prenatal Testing (NIPT) as an optional PDC follow-up service

The California Department of Public Health's Prenatal Screening (PNS) Program will be incorporating cell free fetal DNA testing (Non-invasive Prenatal Testing or NIPT) by November 2013. Under the current PNS Program, if a maternal serum screening result is reported as "Screen Positive", indicating an elevated risk for a chromosomal abnormality or birth defect, then genetic counseling and other follow-up services are provided free of charge at State-approved Prenatal Diagnosis Centers (PDC's).

As of November 4, 2013, the PNS Program will offer NIPT through PDCs as an optional test to all women who are Screen Positive for Down syndrome, trisomy 18, or Large NT (Nuchal Translucency) following biochemical screening. These women, who are at increased risk for a chromosome abnormality, will also have the option of proceeding directly to a diagnostic test such as CVS or amniocentesis. If a NIPT result is negative, no diagnostic tests or further follow-up services will be authorized through the Program. Women with a NIPT negative result in the first trimester will remain eligible for biochemical serum screening in the second trimester, with analyte values reported, but only a risk assessment for neural tube defects (NTDs) and Smith-Lemli-Opitz syndrome (SLOS) reported.

Women with a NIPT positive result through the Program will be authorized to receive confirmatory diagnostic and other follow-up services, and will also remain eligible for serum screening in the second trimester for a risk assessment for NTDs and SLOS.

Patients having NIPT outside the PNS Program are not authorized for further Program PDC services

Women who are Screen Positive for Down syndrome, trisomy 18, or Large NT, and have been drawn for NIPT outside of the PNS Program will not be eligible for PDC follow-up services through the Program.

If the clinician and/or patient chooses NIPT screening outside the Program, this will be considered a choice to have follow-up services outside the Program. If the patient has a "positive" NIPT result outside the Program, additional diagnostic services will have to be obtained using her insurance. The PNS Program will only offer a referral for follow-up services when NIPT obtained outside the Program does not give successful results.

Clinicians who draw patients for both California Prenatal Screening and for NIPT at the same time should make patients aware that this will limit their options for PDC follow-up through the PNS Program.

Other PNS program changes effective October 2013

Women identified to have a fetus with a large NT measurement (≥ 3.5 mm) have a greater than 1 in 5 risk for a chromosome abnormality. These pregnancies are also at risk for congenital heart defects. ACOG guidelines recommend a targeted ultrasound, fetal echocardiogram, or both, for patients with a Large NT, to address the risk for congenital heart defects. The PNS Program's inclusion of fetal echocardiogram for Large NT cases has been problematic. Not all PDC's have offered this service and the later time frame for a fetal echocardiogram makes these cases difficult for Program and PDC staff to track and ensure follow-up. As of October 2013, for patients with Large NT measurements, the PNS Program's follow-up services at PDC's no longer include a fetal echocardiogram. These patients will need to be referred to Pediatric Cardiologists using their own insurance.

In addition, for patients having amniocentesis through the California PNS Program, the Program will no longer cover the cost for amniotic fluid AFP/ACHé analysis, unless the PNS result indicates an increased risk for NTDs. For patients who are Screen Positive for a chromosome abnormality, but Screen Negative for NTDs, almost 99% have negative AF-AFP results following amniocentesis, including the majority of NTD cases found in this group. These NTD cases are identified primarily by ultrasound findings (prepublication Program data).

All of your prenatal screening education materials for patients and clinicians are simply a click away!

www.cdph.ca.gov/pns

- **The Prenatal Screening Calculator**
- **All Patient Booklets**
- **The Provider Handbook**
- **Prenatal Screening Program Information and Updates**

Recent Publications from the California Prenatal Screening Program

The PNS Program appreciates the Outcome of Pregnancy information that is provided by clinicians. The information is used for Program evaluation and development, and for studies relevant to prenatal screening and prenatal care. Some recent publications include:

Currier R, Wu N, Van Meter K, Goldman S, Lorey F, Flessel M. **Integrated and first trimester prenatal screening in California: program implementation and patient choice for follow-up services.** Prenat Diagn 2012 Nov;32(11):1077-83.

Wu N, Platt LD, Greene N, Currier RJ. **Practitioner-specific medians for nuchal translucency to improve first-trimester screening performance.** Obstet Gynecol 2012 Apr;119(4):785-94.

Jelliffe-Pawlowski LL, Shaw GM, Currier RJ, Stevenson, DK, Baer RJ, O'Brodovich HM, et al. **Association of early-preterm birth with abnormal levels of routinely collected first- and second-trimester biomarkers.** Am J Obstet Gynecol 2013 Jun;208(6):492-11.

Second Trimester Test Request Form: CVS Question Now Includes NIPT and Amniocentesis

The second trimester test request form for the California Prenatal Screening Program includes the question: "Has patient had CVS?" If a patient has had CVS prior to second trimester screening, the clinician answers "YES" to this question. For second trimester screening, the Prenatal Screening (PNS) Program then reports analyte values and MoMs, but only reports risk assessment for neural tube defects and SLOS. Risk assessment for Down syndrome and trisomy 18 is not reported.

The CVS question is being expanded to include patients who have had NIPT or amniocentesis prior to second trimester screening. Future versions of the test forms will reflect this change. For current forms on hand, the clinician can mark the CVS question as "YES" and note "NIPT" or "amnio" on the form. The test result mailers to clinicians have been updated to reflect this change. Please contact your regional PNS Coordinator office if you have questions regarding the CVS question.

See enclosed article on Non-invasive Prenatal Testing (NIPT) and the California Prenatal Screening Program.



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ADDRESS SERVICE REQUESTED

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